



## AGXT gene

alanine-glyoxylate aminotransferase

### Normal Function

The *AGXT* gene provides instructions for making an enzyme called alanine-glyoxylate aminotransferase. This enzyme is found in liver cells, specifically within cell structures called peroxisomes. These structures are important for several cellular activities, such as ridding the cell of toxic substances and helping to break down certain fats. In the peroxisome, alanine-glyoxylate aminotransferase converts a compound called glyoxylate to the protein building block (amino acid) glycine.

### Health Conditions Related to Genetic Changes

#### primary hyperoxaluria

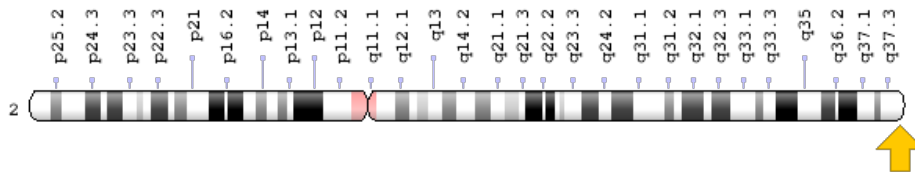
More than 175 mutations in the *AGXT* gene have been found to cause primary hyperoxaluria type 1. This condition is caused by the overproduction of a substance called oxalate. Excess amounts of this substance lead to kidney and bladder stones, which can begin anytime from childhood to early adulthood with kidney disease developing at any age. Deposition of oxalate in multiple other tissues throughout the body (systemic oxalosis) can cause additional health problems.

Most of the *AGXT* gene mutations decrease or eliminate alanine-glyoxylate aminotransferase activity, which impairs the conversion of glyoxylate to glycine. Other mutations cause the enzyme to be misplaced in cells, transporting it to structures called mitochondria instead of to peroxisomes. While the enzyme in the mitochondria retains activity, it cannot access glyoxylate, which is in peroxisomes. All *AGXT* gene mutations result in the accumulation of glyoxylate, which is converted to oxalate instead of glycine. The oxalate is filtered through the kidneys and is either excreted in urine as a waste product or combines with calcium to form calcium oxalate, a hard compound that is the main component of kidney and bladder stones. Increased oxalate levels in the blood can lead to systemic oxalosis, particularly affecting bones and the walls of blood vessels in people with primary hyperoxaluria type 1.

## Chromosomal Location

Cytogenetic Location: 2q37.3, which is the long (q) arm of chromosome 2 at position 37.3

Molecular Location: base pairs 240,868,745 to 240,879,119 on chromosome 2 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- AGT
- AGT1
- AGXT1
- alanine glyoxylate aminotransferase
- alanine-glyoxylate aminotransferase (oxalosis I; hyperoxaluria I; glycolicaciduria; serine-pyruvate aminotransferase)
- alanine-glyoxylate transaminase
- L-alanine: glyoxylate aminotransferase 1
- pyruvate (glyoxylate) aminotransferase
- serine-pyruvate aminotransferase
- serine:pyruvate aminotransferase
- SPAT
- SPT

## Additional Information & Resources

### GeneReviews

- Primary Hyperoxaluria Type 1  
<https://www.ncbi.nlm.nih.gov/books/NBK1283>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28AGXT%5BTIAB%5D%29+OR+%28alanine-glyoxylate+aminotransferase%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5BIa%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>

### OMIM

- ALANINE-GLYOXYLATE AMINOTRANSFERASE  
<http://omim.org/entry/604285>

### Research Resources

- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=AGXT%5Bgene%5D>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=341](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=341)
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/189>
- UniProt  
<http://www.uniprot.org/uniprot/P21549>

### **Sources for This Summary**

- Cochat P, Rumsby G. Primary hyperoxaluria. N Engl J Med. 2013 Aug 15;369(7):649-58. doi: 10.1056/NEJMra1301564. Review. Erratum in: N Engl J Med. 2013 Nov 28;369(22):2168.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/23944302>
- GeneReview: Primary Hyperoxaluria Type 1  
<https://www.ncbi.nlm.nih.gov/books/NBK1283>
- Hopp K, Cogal AG, Bergstralh EJ, Seide BM, Olson JB, Meek AM, Lieske JC, Milliner DS, Harris PC; Rare Kidney Stone Consortium. Phenotype-Genotype Correlations and Estimated Carrier Frequencies of Primary Hyperoxaluria. J Am Soc Nephrol. 2015 Oct;26(10):2559-70. doi: 10.1681/ASN.2014070698. Epub 2015 Feb 2.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/25644115>  
*Free article on PubMed Central:* <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4587693/>
- Williams EL, Acquaviva C, Amoroso A, Chevalier F, Coulter-Mackie M, Monico CG, Giachino D, Owen T, Robbiano A, Salido E, Waterham H, Rumsby G. Primary hyperoxaluria type 1: update and additional mutation analysis of the AGXT gene. Hum Mutat. 2009 Jun;30(6):910-7. doi: 10.1002/humu.21021. Review.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/19479957>

---

Reprinted from Genetics Home Reference:  
<https://ghr.nlm.nih.gov/gene/AGXT>

Reviewed: December 2015

Published: March 21, 2017

Lister Hill National Center for Biomedical Communications  
U.S. National Library of Medicine  
National Institutes of Health  
Department of Health & Human Services